

<!--StartFragment-->RESULT 7

MYPR_HUMAN

ID MYPR_HUMAN Reviewed; 277 AA.
 AC P60201; P04400; P06905; Q502Y1;
 DT 01-JAN-1988, integrated into UniProtKB/Swiss-Prot.
 DT 23-JAN-2007, sequence version 2.
 DT 08-APR-2008, entry version 56.
 DE Myelin proteolipid protein (PLP) (Lipophilin).
 GN Name=PLP1; Synonyms=PLP;
 OS Homo sapiens (Human).
 OC Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 OC Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
 OC Catarrhini; Hominidae; Homo.
 OX NCBI_TaxID=9606;
 RN [1]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA].
 RX MEDLINE=87092337; PubMed=3467339;
 RA Diehl H.-J., Schaich M., Budzinski R.-M., Stoffel W.;
 RT "Individual exons encode the integral membrane domains of human myelin
 RT proteolipid protein.";
 RL Proc. Natl. Acad. Sci. U.S.A. 83:9807-9811(1986).
 RN [2]
 RP NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM DM-20).
 RX MEDLINE=87298492; PubMed=2441695; DOI=10.1016/0006-291X(87)90580-8;
 RA Simons R., Alon N., Riordan J.R.;
 RT "Human myelin DM-20 proteolipid protein deletion defined by cDNA
 RT sequence.";
 RL Biochem. Biophys. Res. Commun. 146:666-671(1987).
 RN [3]
 RP NUCLEOTIDE SEQUENCE [MRNA], AND VARIANT PMD ARG-163.
 RX MEDLINE=90046751; PubMed=2479017;
 RA Hudson L.D., Puckett C., Berndt J., Chan J., Gencic S.;
 RT "Mutation of the proteolipid protein gene PLP in a human X chromosome-
 RT linked myelin disorder.";
 RL Proc. Natl. Acad. Sci. U.S.A. 86:8128-8131(1989).
 RN [4]
 RP NUCLEOTIDE SEQUENCE [LARGE SCALE GENOMIC DNA].
 RX PubMed=15772651; DOI=10.1038/nature03440;
 RA Ross M.T., Grafham D.V., Coffey A.J., Scherer S., McLay K., Muzny D.,
 RA Platzer M., Howell G.R., Burrows C., Bird C.P., Frankish A.,
 RA Lovell F.L., Howe K.L., Ashurst J.L., Fulton R.S., Sudbrak R., Wen G.,
 RA Jones M.C., Hurler M.E., Andrews T.D., Scott C.E., Searle S.,
 RA Ramser J., Whittaker A., Deadman R., Carter N.P., Hunt S.E., Chen R.,
 RA Cree A., Gunaratne P., Havlak P., Hodgson A., Metzker M.L.,
 RA Richards S., Scott G., Steffen D., Sodergren E., Wheeler D.A.,
 RA Worley K.C., Ainscough R., Ambrose K.D., Ansari-Lari M.A., Aradhya S.,
 RA Ashwell R.I., Babbage A.K., Bagguley C.L., Ballabio A., Banerjee R.,
 RA Barker G.E., Barlow K.F., Barrett I.P., Bates K.N., Beare D.M.,
 RA Beasley H., Beasley O., Beck A., Bethel G., Blechschmidt K., Brady N.,
 RA Bray-Allen S., Bridgeman A.M., Brown A.J., Brown M.J., Bonnin D.,
 RA Bruford E.A., Buhay C., Burch P., Burford D., Burgess J., Burrill W.,
 RA Burton J., Bye J.M., Carder C., Carrel L., Chako J., Chapman J.C.,
 RA Chavez D., Chen E., Chen G., Chen Y., Chen Z., Chinault C.,
 RA Ciccodicola A., Clark S.Y., Clarke G., Clee C.M., Clegg S.,
 RA Clerc-Blankenburg K., Clifford K., Copley V., Cole C.G., Conquer J.S.,
 RA Corby N., Connor R.E., David R., Davies J., Davis C., Davis J.,
 RA Delgado O., Deshazo D., Dhami P., Ding Y., Dinh H., Dodsworth S.,
 RA Draper H., Dugan-Rocha S., Dunham A., Dunn M., Durbin K.J., Dutta I.,
 RA Eades T., Ellwood M., Emery-Cohen A., Errington H., Evans K.L.,
 RA Faulkner L., Francis F., Frankland J., Fraser A.E., Galgoczy P.,
 RA Gilbert J., Gill R., Gloeckner G., Gregory S.G., Gribble S.,

RA Griffiths C., Grocock R., Gu Y., Gwilliam R., Hamilton C., Hart E.A.,
 RA Hawes A., Heath P.D., Heitmann K., Hennig S., Hernandez J.,
 RA Hinzmann B., Ho S., Hoffs M., Howden P.J., Huckle E.J., Hume J.,
 RA Hunt P.J., Hunt A.R., Isherwood J., Jacob L., Johnson D., Jones S.,
 RA de Jong P.J., Joseph S.S., Keenan S., Kelly S., Kershaw J.K., Khan Z.,
 RA Kioschis P., Klages S., Knights A.J., Kosiura A., Kovar-Smith C.,
 RA Laird G.K., Langford C., Lawlor S., Leversha M., Lewis L., Liu W.,
 RA Lloyd C., Lloyd D.M., Loulseged H., Loveland J.E., Lovell J.D.,
 RA Lozado R., Lu J., Lyne R., Ma J., Maheshwari M., Matthews L.H.,
 RA McDowall J., McLaren S., McMurray A., Meidl P., Meitinger T.,
 RA Milne S., Miner G., Mistry S.L., Morgan M., Morris S., Mueller I.,
 RA Mullikin J.C., Nguyen N., Nordsiek G., Nyakatura G., O'dell C.N.,
 RA Okwuonu G., Palmer S., Pandian R., Parker D., Parrish J.,
 RA Pasternak S., Patel D., Pearce A.V., Pearson D.M., Pelan S.E.,
 RA Perez L., Porter K.M., Ramsey Y., Reichwald K., Rhodes S.,
 RA Ridler K.A., Schlessinger D., Schueler M.G., Sehra H.K.,
 RA Shaw-Smith C., Shen H., Sheridan E.M., Shownkeen R., Skuce C.D.,
 RA Smith M.L., Sotheran E.C., Steingruber H.E., Steward C.A., Storey R.,
 RA Swann R.M., Swarbreck D., Tabor P.E., Taudien S., Taylor T.,
 RA Teague B., Thomas K., Thorpe A., Timms K., Tracey A., Trevanion S.,
 RA Tromans A.C., d'Urso M., Verduzco D., Villasana D., Waldron L.,
 RA Wall M., Wang Q., Warren J., Warry G.L., Wei X., West A.,
 RA Whitehead S.L., Whiteley M.N., Wilkinson J.E., Willey D.L.,
 RA Williams G., Williams L., Williamson A., Williamson H., Wilming L.,
 RA Woodmansey R.L., Wray P.W., Yen J., Zhang J., Zhou J., Zoghbi H.,
 RA Zorilla S., Buck D., Reinhardt R., Poustka A., Rosenthal A.,
 RA Lehrach H., Meindl A., Minx P.J., Hillier L.W., Willard H.F.,
 RA Wilson R.K., Waterston R.H., Rice C.M., Vaudin M., Coulson A.,
 RA Nelson D.L., Weinstock G., Sulston J.E., Durbin R.M., Hubbard T.,
 RA Gibbs R.A., Beck S., Rogers J., Bentley D.R.;
 RT "The DNA sequence of the human X chromosome.";
 RL Nature 434:325-337(2005).
 RN [5]
 RP NUCLEOTIDE SEQUENCE [LARGE SCALE MRNA] (ISOFORMS 1 AND DM-20).
 RC TISSUE=Spinal cord, and Uterus;
 RX PubMed=15489334; DOI=10.1101/gr.2596504;
 RG The MGC Project Team;
 RT "The status, quality, and expansion of the NIH full-length cDNA
 RT project: the Mammalian Gene Collection (MGC).";
 RL Genome Res. 14:2121-2127(2004).
 RN [6]
 RP PROTEIN SEQUENCE OF 2-277.
 RX MEDLINE=86000127; PubMed=4041237;
 RA Stoffel W., Giersiefen H., Hillen H., Schroeder W., Tunggal B.;
 RT "Amino-acid sequence of human and bovine brain myelin proteolipid
 RT protein (lipophilin) is completely conserved.";
 RL Biol. Chem. Hoppe-Seyler 366:627-635(1985).
 RN [7]
 RP NUCLEOTIDE SEQUENCE.
 RC TISSUE=Spinal cord;
 RX MEDLINE=88141333; PubMed=2449536;
 RA Kronquist K.E., Crandall B.F., Macklin W.B., Campagnoni A.T.;
 RT "Expression of myelin proteins in the developing human spinal cord:
 RT cloning and sequencing of human proteolipid protein cDNA.";
 RL J. Neurosci. Res. 18:395-401(1987).
 RN [8]
 RP VARIANT PMD SER-217.
 RX MEDLINE=89371750; PubMed=2773936;
 RA Gencic S., Abuelo D., Ambler M., Hudson L.D.;
 RT "Pelizaeus-Merzbacher disease: an X-linked neurologic disorder of
 RT myelin metabolism with a novel mutation in the gene encoding

RT proteolipid protein.";
 RL Am. J. Hum. Genet. 45:435-442(1989).
 RN [9]
 RP VARIANT PMD LEU-15.
 RX MEDLINE=90083280; PubMed=2480601;
 RA Trofatter J., Dlouhy S.R., Demyer W., Conneally P.M., Hodes M.E.;
 RT "Pelizaeus-Merzbacher disease: tight linkage to proteolipid protein
 RT gene exon variant.";
 RL Proc. Natl. Acad. Sci. U.S.A. 86:9427-9430(1989).
 RN [10]
 RP VARIANT PMD ILE-156.
 RX MEDLINE=91214553; PubMed=1708672;
 RA Weimbs T., Dick T., Stoffel W., Boltshauser E.;
 RT "A point mutation at the X-chromosomal proteolipid protein locus in
 RT Pelizaeus-Merzbacher disease leads to disruption of myelinogenesis.";
 RL Biol. Chem. Hoppe-Seyler 371:1175-1183(1990).
 RN [11]
 RP VARIANT PMD ILE-156.
 RX MEDLINE=91189235; PubMed=1707231;
 RA Pratt V.M., Trofatter J.A., Schinzel A., Dlouhy S.R., Conneally P.M.,
 RA Hodes M.E.;
 RT "A new mutation in the proteolipid protein (PLP) gene in a German
 RT family with Pelizaeus-Merzbacher disease.";
 RL Am. J. Med. Genet. 38:136-139(1991).
 RN [12]
 RP VARIANT PMD PHE-219.
 RX MEDLINE=91352028; PubMed=1715570;
 RA Pham-Dinh D., Popot J.-L., Bosepflug-Tanguy O., Landrieu P.,
 RA Deleuze P., Boue J., Jolles P., Dautigny A.;
 RT "Pelizaeus-Merzbacher disease: a valine to phenylalanine point
 RT mutation in a putative extracellular loop of myelin proteolipid.";
 RL Proc. Natl. Acad. Sci. U.S.A. 88:7562-7566(1991).
 RN [13]
 RP VARIANTS PMD ARG-74 AND HIS-203.
 RX MEDLINE=92303562; PubMed=1376966;
 RA Doll R., Natowicz M.R., Schiffmann R., Smith F.I.;
 RT "Molecular diagnostics for myelin proteolipid protein gene mutations
 RT in Pelizaeus-Merzbacher disease.";
 RL Am. J. Hum. Genet. 51:161-169(1992).
 RN [14]
 RP VARIANTS PMD PRO-182 AND PRO-224.
 RX MEDLINE=93035344; PubMed=1384324;
 RA Strautnieks S., Rutland P., Winter R.M., Baraitser M., Malcolm S.;
 RT "Pelizaeus-Merzbacher disease: detection of mutations Thr181-->Pro and
 RT Leu223-->Pro in the proteolipid protein gene, and prenatal
 RT diagnosis.";
 RL Am. J. Hum. Genet. 51:871-878(1992).
 RN [15]
 RP VARIANT PMD GLU-166.
 RX MEDLINE=93276877; PubMed=7684886;
 RA Pratt V.M., Kiefer J.R., Lahdetie J., Schleutker J., Hodes M.E.,
 RA Dlouhy S.R.;
 RT "Linkage of a new mutation in the proteolipid protein (PLP) gene to
 RT Pelizaeus-Merzbacher disease (PMD) in a large Finnish kindred.";
 RL Am. J. Hum. Genet. 52:1053-1056(1993).
 RN [16]
 RP VARIANT PMD SER-217.
 RX MEDLINE=93176327; PubMed=7679906;
 RA Otterbach B., Stoffel W., Ramaekers V.;
 RT "A novel mutation in the proteolipid protein gene leading to
 RT Pelizaeus-Merzbacher disease.";

RL Biol. Chem. Hoppe-Seyler 374:75-83(1993).
RN [17]
RP VARIANT PMD CYS-221.
RX MEDLINE=93258343; PubMed=7683951; DOI=10.1093/hmg/2.1.19;
RA Iwaki A., Muramoto T., Iwaki I., Furumi H., Dario-Deleon M.L.,
RA Tateishi J., Fukumaki Y.;
RT "A missense mutation in the proteolipid protein gene responsible for
RT Pelizaeus-Merzbacher disease in a Japanese family.";
RL Hum. Mol. Genet. 2:19-22(1993).
RN [18]
RP VARIANT PMD SER-216.
RX MEDLINE=94311323; PubMed=8037216;
RA Pratt V.M., Boyadjiev S., Dlouhy S.R., Silver K., der Kaloustian V.M.,
RA Hodes M.E.;
RT "Comparison of statistics for candidate-gene association studies using
RT cases and parents.";
RL Am. J. Hum. Genet. 55:402-404(1994).
RN [19]
RP VARIANT SPG2 TYR-140.

Query Match 87.5%; Score 70; DB 1; Length 277;
Best Local Similarity 84.6%; Pred. No. 0.0095;
Matches 11; Conservative 1; Mismatches 1; Indels 0; Gaps 0;

Qy 1 HSLGKWLGHPNKF 13
| |||||:|
Db 140 HCLGKWLGHDPKF 152
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